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Mutational analysis of *CTLA-4* gene and HLA associations in SLE patients from Lahore-Pakistan

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Abstract

Systemic Lupus Erythematosus is one of the classic examples of autoimmune diseases among human beings and is a rare disease in Pakistani population. In this study, we had focused on the mutational analysis of *CTLA-4* gene and HLA associations in SLE patients from Lahore-Pakistan. A total of 122 individuals were enrolled in this study. Of the 122, 61 were the SLE patients who fulfilled revised ACR criteria and 61 were the healthy controls. Mean age of SLE patients at diagnosis was 30.35 ± 1.687 years (Range: 12-68 years). Mutational analysis of *CTLA4* gene exon 1 was performed because most of the polymorphisms were reported in this region with reference to SLE. *CTLA4* gene exon 1 was amplified by Step-down PCR technique in order to get the band of our interest that is of 354 bp. Extracted DNA band of 354 bp was sequenced to analyze mutations in the exon-1 of *CTLA4* gene. No *CTLA-4* exon 1 polymorphism was found in SLE patients after DNA sequence analysis. Nucleotide sequence was further translated in to proteins and found to be resembled with Chinese SLE population. The distribution of HLA-A, -B, -DRB1 alleles was analyzed in SLE patients with various disease phenotypes. Certain MHC genes were identified to be involved in the development of SLE like a significant increase was observed in the frequency of HLA-A*01, -A*03, -A*11, -A*23, -A*26, -A*69, HLA-B*27, -B*40, -B*49, -B*51, -B*52, -B*53, -B*54, -B*95, HLA-DRBI*01, -DRBI*03, -DRBI*11, -DRBI*14, -DRBI*16 alleles among total SLE patients.

Key Words: Systemic Lupus Erythematosus, Human Leukocyte Antigens, Major Histocompatibility Complex, Cytotoxic T lymphocyte Antigens.

Biography

Nageen Hussain has completed her Ph.D in 2012 at the age of 31 years from University of the Punjab. Currently she is working as an Assistant Professor in the Department of Microbiology and Molecular Genetics, University of the Punjab, Lahore-Pakistan. She has joined the same department in 2005 as a Lecturer. She has taught basic immunology, cellular and molecular immunology, microbial diversity and bacterial metabolism. She has supervised five BS Hons. and two M.Sc. research students. Now she is supervising 1 BS Hons., 1 M.Sc. and 1 M.Phil research students. She has completed 6 research projects awarded by the Punjab University.

Nageen Hussain has published more than 15 papers in reputed National and International journals which speaks the merit of her work. She is serving as an editorial board member of WASET Scientific Committee and also as a Co-Editor of "International Journal of Health Sciences". She has presented her research work not only at the National level but also Internationally in Japan (14th international congress of Immunology held in Kobe-Japan from August 22-27, 2010), Amsterdam (International Conference on Bioinformatics and Biomedical Engineering, Amsterdam, The Netherlands, 13-15 July, 2011) as well as in Switzerland (International Conference on Environmental, Biological, social sciences and Engineering, Lucerne, Switzerland, 15-16 October, 2012). She has also participated in the 8th FIMSA advanced Immunology course, 1-5 March, 2006.

She has a self learning ability in research and does her work with modification and amendments according to her Research nature/requirements. She is dutiful cooperative, flexible and punctual. Her attitude towards administrative works, arranging seminars and workshops is also very positive and willing. She has the ability to present her research work confidently and with a high degree of diligence.